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THOMSEN'S DISEASE (MYOTONIA CONGENITA.)

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THE history of this peculiar affection has been so lately and so thoroughly given by Erb[1] in his monograph upon this subject, that it would be superfluous to repeat the same here. Erb has examined all cases reported as cases of Thomsen's disease, and has carefully sifted them. He divides them into three classes; the first containing only purely typical cases, the second those which are more or less dubious, those which do not agree in every important point, but which at the same time show a great similarity to pure cases, and the third those which, although showing the principal symptoms, the "myotonic disorder of the muscles," still, on account of their complication with other symptoms, cannot be considered as true cases of Thomsen's disease, but are probably cases of central affection.

It will not be amiss to quote the following short definition of the affection from Erb. "I understand by the typi-

[1] Erb. W., "Die Thomsensche Krankheit (Myotonia Congenita)." Leipzig, 1886.

cal form of Thomsen's disease those forms of disease which generally, under the influence of an hereditary or family noxiousness, already in very early youth show that disorder of movements which later becomes more pronounced. The occurrence after a period of rest, and during movements, of tension and stiffness of the muscles up to complete inhibition of movement, gradual dissolution of this tension by means of continued movement, until entire relief occurs. Prolongation of the tonic contractions, produced by energetic action of the will, and inability to voluntarily relax the muscles quickly. In connection with this a remarkably strong, hypertrophic development of the voluntary muscles, in direct contrast to their relatively small power. The muscles also showing certain characteristic changes in their mechanical and electrical excitability, particularly the prolongation of artificially produced contractions; all other functions being perfectly normal, the nervous system especially not showing any other disorder."

Among the purely typical cases Erb classes those occurring in Thomsen's own family (twenty-two or twenty-three; two of which, those of Dr. Thomsen himself and one of his sons, only having been described, and even these without any objective examination), cases described by Leyden (one), Seeligmuller (two), Bernhardt (three), Peters (one), Strümpell (one), Petrone (one), Westphal (two), Ballet and Marie (one), Weichmann (two), Rieder (one), Knud Pontoppidan (four), Romain Vigouroux (one), Pitres and Dallidet (one), Eulenburg and Melchert (four), and Fischer (one), making in all, with Erb's cases, twenty-eight. Under the heading of doubtful cases he classes eleven.

The only case in the English language referred to is that of Engel and this case is classed as dubious.

Engel's case [1] bears in many respects a certain resemblance to a true case of Thomsen's disease, but it also

[1] H. Engel. "A Case of Thomsen's disease. A form of Paresis of Motion accompanied by muscular hypertrophy." Phila. Med. Times, Sept. 8th, 1883.

differs in so many important particulars that it must be classed under some other heading. The points which show its diversity are the following:

The affection occurred suddenly in a youth 17 years of age in consequence of fright, caused by lightning striking a tree near the place where he was standing. The electro-muscular contractility is increased in the legs alone. Of this it is said that the response is quicker and the contraction stronger than in the other muscles, but not a word is said of prolongation or tonicity of the contractions. The muscular sense is impaired. There was present a mild foot clonus. A gradually increasing resistance was present to passive motion.

In the morning after getting up he has no difficulty whatever. The arms are not at all affected, and finally the description of the patient's manner of walking and falling differs entirely from that in Thomsen's disease. Engel says, "When told, after he had been standing erect for a long time, to walk, he lifted his left leg with great difficulty, and when the front part of the foot touched the floor the thigh was bent inwardly at the hip and the leg bent backward at the knee. Then the other leg underwent the same peculiar motion, and while this was going on, the upper part of the body swayed to and fro, a symptom which became worse the moment the other foot touched the ground; then, with steadily increasing difficulty, the same process was repeated at the next step, the swaying of the body increasing still more, and at the commencement of the third step, the left foot gave way, gliding under the right leg, while the opposite took place with the right foot, so that the patient suddenly fell forward," etc., and further on, "almost the same phenomenon happened after he had been sitting awhile, only that then the upper part of the body swayed to and fro, from the moment he arose, and before he made the first attempt at locomotion," etc.

In various American journals, references are found to "Hamilton's cases of Thomsen's disease." The only article to which these references can allude is that published

by Dr. Allen McLane Hamilton, of this city, in the New York *Medical Record*, p. 85, 1886, and entitled " A Consideration of the Thomsen Symptom Complex with Reference to a new form of Paralysis Agitans." It is clear from a perusal of this article that Dr. Hamilton does not regard these cases as genuine cases of Thomsen's disease, but as cases of central trouble presenting certain symptoms of the former affection. Dr. Hamilton, in a letter to me, says that this interpretation of his article is in accordance with his views.

The case which I desire to describe was referred to me on Oct. 28th, 1886, by Dr. A. C. Bucklin, of this city, whom he consulted in reference to his eyes, and was shown at the meeting of the New York Neurological Society, Nov. 2d, 1886.

George H. Glenn, from Cape Vincent, Jefferson County, N. Y., æt. 24, painter.

The family history is unsatisfactory, and nothing important from an hereditary point of view is discoverable. He does not remember his parents, who both died when he was a child. He never had any brothers or sisters, and his only living relative is an aunt who is perfectly healthy. He thinks that his maternal grandfather had some nervous trouble. He is unable to assign any cause for his infirmity. The affection has existed as long as he can remember, and he is unable to recollect any time when he was perfectly well. He remembers distinctly that he could not participate in the out-door games of his comrades on account of stiffness of his muscles. He could not run like other boys and was debarred from joining in athletic sports. He attempted to learn to play upon the organ, but was unable to make any progress on account of the clumsiness of his hands. What he particularly noticed was that all of his muscles were stiff, and that the execution of every movement was impaired. This was particularly noticeable after periods of prolonged rest. The first few movements always decidedly increased this stiffness, but then it gradually diminished and finally entirely disappeared, so that a perfectly free use of the muscles set in. This stiffness always reappeared after a pause or rest. All the voluntary muscles were affected, those of the upper as well as the lower extremities. Also the muscles of his face are involved. When he attempts to whistle he finds difficulty in doing so. In mastication, the muscles become stiff and rigid, and he is affected, as it were, with a temporary trismus. This stiffness was also noticeable in closing his eyes. Only such muscles or groups of muscles as were at the time employed would become thus affected, all the inactive mus-

cles remained perfectly lax. This tonic contraction of the muscles was, as far as he remembers, never accompanied by any pain. He thinks that he was decidedly worse when a boy than at present. If in running he stubbed his toe against any hard protruding object, all the muscles of his body would stiffen up like a board and he would fall, being unable to rise for several seconds. This does not occur now. At about the age of fourteen, he began to learn his trade, and he attributes his improvement to being continually at work, for, he says, "moving around and working limbers me up," and, "if I did not work now, but just kept quiet, I would be as bad as formerly." There were always periods of time during which he was better than at others. Mental influence did not have any effect upon his bodily condition. He says that he does not grow worse when thinking of it, or when excited. I desire, however, here to remark that when I presented him before the members of the New York Neurological Society, he was decidedly worse than I had ever seen him before, and I attributed this circumstance to the mental excitement caused by this presentation, although he himself would not acknowledge the correctness of my explanation. He also noticed that his muscles were large in comparison to other boys', and was astonished and chagrined to find that he had so little force in them. He also noticed a difficulty in bringing his eyes back to a certain point, and frequently after looking at some object situated far to one side for a short space of time, and then trying to look at something in front of him, he found that for a few seconds he would either not be able to see at all, or he would see double, but that by a shake of the head he would "bring his eyes around all right." About two years ago, after having been considerably troubled in this way and upon regarding himself in the mirror, he found that his right eye was "turned in." In every other way he was perfectly healthy.

Status præsens.—Patient is a person of medium height and strong bony structure; he has not a thick panniculus adiposus. His skin is of normal appearance. The shape of his body is normal, except in the exceedingly strong development of nearly all of the muscles. This at once attracts attention, being noticeable even with his clothes on. When undressed he looks decidedly athletic. Particularly strongly developed are the muscles of the leg and thigh, those of the gluteal region, and those of the arm and forearm. The muscles of the shoulders and neck are also extraordinarily prominent. The entire appearance, excepting that of size, is, when undressed, herculean. The face, however, does not show any of this hypertrophy. The following measurements will give an idea as to the size of the limbs: Right calf, 15 inches; left, 15½ inches. Right thigh, 7 inches above patella, 23 inches. Left thigh at the same place, 23 inches. Forearm above, right, 11 inches; left, 10½ inches. Arms, middle (over the biceps), extended, right, 12 inches; left 11½ inches; flexed, right, 14 inches; left, 13½ inches. Thorax over xiphoid process, 35 inches. Thorax over nipples, 36½ inches.

The heart, lungs, and abdominal organs are normal in every particular. Temperature and respiration also normal. Respiration when voluntarily increased does not show any difficulty; but expiration after a forced inspiration is occasionally difficult and protracted. The urine is normal. Joints freely movable. The nervous system was found normal in every way. Psychical condition, intelligence, and memory appeared to me to be normal. The patient himself says that his memory is poor, but I was not able to satisfy myself upon this point. Hearing good. Eyes and speech present certain anomalies to which I will again recur.

The sensibility of the skin over the entire body is normal. No vasomotor changes. No affections of the bladder. Cremaster, abdominal, and plantar reflexes present. No foot clonus. Tendon reflexes can be obtained from the patellar and triceps tendons, and occasionally also from the tendo Achilles. The patellar tendon reflex presents certain curious features which must be specially mentioned. Sometimes, especially if the reflex has not been tested for a long period of time, the first blow produces a decidedly exaggerated contraction of the muscle. This reflex contraction then diminishes with each successive blow, becomes normal, less than normal, and at times entirely disappears. The exaggeration is not always demonstrable, but the gradual diminution from the normal to total disappearance can generally be easily obtained; even to this, however, there are exceptions. All in all, the condition of this as well as of the other tendon reflexes was found to vary at various times.

The *muscular system* is the part in which the most noticeable changes are observed. Besides the great hypertrophy, the most apparent symptom is a certain stiffness and slowness of all movements which are executed after a period of rest. The movements which demonstrate this most forcibly are those of flexion and extension, and the patient himself notices the increasing stiffness as it is coming on, but before it is fully developed. If then the same movements through which the stiffness has been produced are repeatedly executed, this stiffness is gradually lost, and the patient can execute the previously inhibited movement without any trouble. If the patient is asked to execute any forcible movement such as forced flexion of the hand, of the arm, or of the leg, a tonic contraction of the exerted muscles takes place which lasts for some time after all influence of the will has disappeared. This tonic contraction is so great that for the time being all antagonistic movements are rendered impossible. This same phenomenon is observed after forcible closure of the eyelids, when he is unable to open them until after the lapse of many seconds. The power of the muscles stands in marked contrast to their enormous development, for it is easily shown to be decidedly diminished. Examination with a dynamometer is interesting, for it not only demonstrates the reduction of muscular force, but also shows the characteristic inability to relax the muscles which have been employed in the

pressure, as shown by the slowness and awkwardness with which the dynamometer is released from the grasp.

This highly interesting and pathognomonic symptom is present in all of the voluntary muscles of the body. The muscles of the lips are sometimes affected when the patient attempts to whistle or to forcibly pronounce a labial as P or B. In eating, that is to say in swallowing, it is occasionally difficult to get the bolus down; "it sticks in the throat." The tongue is also sometimes affected when he first begins to talk; for the last two statements I have taken the patient's word, I myself have not observed them. The movements of the eyes are also sometimes affected. Upon the right eye there is strabismus; this and other interesting features connected with the eyes are explained in the following extracts of a long letter from Dr. C. A. Bucklin, of this city, by whom the patient was kindly sent me. Dr. Bucklin, after carefully going over a variety of conditions which are likely to produce convergent strabismus, for this patient is affected with the convergent form, says:

"I am certain that no case of strabismus ever developed in a person who could see distinctly and with comfort at all distances with both eyes. This patient could not see at all distances with both eyes, because he was prevented from co-ordinating his visual axes by the tonic spasms of his internal recti muscles. For many years he would see double at every sudden movement that he made with his eyes; by closing his eyes for an instant, then upon opening them and giving his head a sudden rotating movement in both directions, he would find a point where he could fix with both eyes and see distinctly. As years went on he discovered that it was more and more difficult to make the diplopia disappear, owing to the fact of his internal recti muscles being so much stronger than the other ocular muscles and their also being more frequently affected by this spasm.

"These muscles finally began to shorten, so that he could only occasionally relax them and dispel the double vision. One day he accidentally discovered that he could voluntarily ignore the image of the deviating eye; from this moment he made only occasional attempts to see with both eyes, owing to the extreme difficulty of doing so. When an object is brought sufficiently near to make it convenient for him to see with both eyes, he fixes binocularly, and is perfectly conscious of seeing with both eyes. When the fixed object is suddenly removed two feet away he gives up fixing with both eyes and promptly declares that the deviating eye is no longer of any use to him. It is an interesting fact that he is conscious of the change from monocular to binocular vision.

"The vision of each eye is $\frac{20}{30}$ on a bright day; it is not improved by any form of lenses. If he is asked to fix an object at the extreme right or the extreme left, some seconds elapse before he can fix upon an object in any other position. Both pupils react to light rather sluggishly. The deviating eye has a slightly larger pupil than the other eye, which reacts to light still more sluggishly. In a dark room he sees more distinctly with the deviating

eye than with the other one. Under these circumstances he frequently uses the deviating eye instead of the fixing one.

"The fundus is normal.

"The mobility of both eyes inward is abnormally great, their mobility in all other directions being normal. It is thus seen that Thomsen's disease may simply be another cause to disturb the natural relations existing between fixation and accommodation."

Fibrillary tremor in the muscles is sometimes present, but, besides this, I have twice noticed, once in the deltoid and once in the biceps, a lifting up and vermiform movement of entire groups of muscular fibres. There is also present a certain amount of muscular restlessness, so that the patient finds it difficult to sit quiet very long without moving about in his chair. Compression of the nerves or arteries does not produce any muscular contractions. The hypertrophied muscles are elastic and full to the touch, but there is nothing to be found which could be looked upon as an induration; the impression which they make upon the observer's hand is entirely different from the muscles in pseudo-hypertrophic paralysis. The joints are freely movable and passive movement does not produce any contractions of the muscles.

Mechanical excitability.—The nerves themselves do not show any abnormal mechanical excitability; the muscles, however, show a marked exaltation in this respect. I have not been able, as Erb has in his cases, to produce any contractions by simple pressure with the fingers or by rolling the muscle between the hands, but the application of an Esmarch bandage (applied for the excision of a piece of the quadriceps femoris), produced violent tonic contractions of the muscles of the entire leg. If, however, the muscle is given a sharp blow, as with a percussion hammer, this mechanical hyper-excitability becomes very manifest. When such a blow is given, a deep groove is formed in the muscle, running for some distance from the point of excitation; it seems as if the parts on either side of the irritated portion rise up and leave the furrow between them. The rising up of the irritated fibres like a thick cord, as Erb describes it, does not occur.

Smaller muscles, the interossei for instance, contract as a whole, and the contraction lasts for some time after the excitation has ceased. The length of time which the first described groove persists is something remarkable, and I have repeatedly seen it last from twenty to forty seconds. All of the voluntary muscles which were examined clearly showed this phenomenon, some to a greater, others to a lesser extent.

The *electrical examination* was made with great care and each experiment frequently repeated. Erb's cases were taken as guides.

Motor nerves, faradic excitability. Erb's results were fully corroborated. The experiments were performed over the accessory, ulnar and peroneal nerves. Very weak currents that, is, the minimum amount of current requisite to produce a contraction, brought about contractions which were not prolonged. Strong

currents produced contractions lasting from fifteen to twenty-six seconds after removal of the current. Single opening shocks never, even with the strongest currents, produce any prolonged contractions, but only short quick ones.

The *galvanic excitability* of the nerves showed nothing abnormal; tonic prolonged contractions of the muscles, as seen in consequence of their direct excitation, do not occur. Neither is there any reversal of the normal formula.

Muscles. Faradic excitability is very marked, minimum currents produced normal contractions, without any prolongation. Strong excitation produces contractions which last from twenty to thirty seconds after the current is broken. Single opening shocks uniformly produce only a short quick contraction, no matter how strong a current is employed.

Galvanic excitability was also found well pronounced. The muscles are very easily excited. They react only to closure contraction. The excitation is not always confined to the muscle acted upon, but seems to spread easily to contiguous muscles, so that entire groups are brought into contraction. Qualitatively what impressed me most was that the K.C.C. and An.C.C. do not seem to bear any fixed relationship to each other. This is very manifest. In most muscles the K.C.C. is obtained first and is followed at once, with only a very slight increase of current, by the An.C.C., but in some muscles they are both obtained with one and the same strength of current, and in others again the An. C.C. precedes the K.C.C. Besides this, the slowness with which the contractions take place and their long persistence is very noticeable. With fairly strong currents this slowness of contraction is shown by the gradual appearance of a deep groove in the muscle, which, if the electrode is moved along, can be moved with the electrode. If now the current be broken, the long persistence of the contraction becomes apparent, and this persistence is always in direct proportion to the strength of the current employed.

It is hardly necessary to give the figures as obtained from the various electrical tests; they do not differ materially from Erb's. *Rhythmical wave-like* or *undulatory contractions*, as described by Erb, I have at no time been able to obtain; but what I have repeatedly seen, and this has been verified by others who were thoroughly impartial, was a tonic contraction of the muscular groups, those at the kathode becoming first affected and finally those at the anode becoming involved, until the entire extremity was in a state of tonic contraction.

This successive implication of the various groups was very evident with different strengths of the current varying from 10 to 20 m.a. The stronger the current the more marked were the contractions. In the arm the kathodic-anodic contraction was always very plain. In the thigh the direction was not so evident.

The nerves of the arm, the radial, ulnar, and median, seemed to be particularly well adapted for the study of the difference between direct and indirect excitation. One and the same current in the

first case always producing short quick contractions and in the second slow, tonic, lasting ones.

All of the changes above described were found to be present in every muscle examined; some muscles, however, showed them more, others less distinctly. They were at all times most marked in the upper extremities.

The patient remained under observation until Nov. 5th, when he left New York to return home. Since then I have had two letters from him, a few sentences of which may be of interest.

In the first letter, dated November 7th, he says:

"I think the weather has something to do with my case. When I left New York, I felt all right. It was, as you know, a fine day. Well, a little after dark I got very restless; my legs began to ache and got very stiff, and when I got off the train at Watertown it was raining, so you see it must have been the change of atmosphere that affected me, and it always does. I forgot to tell you, doctor, that I had some trouble when born. I was delivered with instruments." The next letter is dated December 12th. In this he says: "I always feel better in winter; I will tell you when I begin to grow worse; it is generally about the breaking up of winter. We live up-stairs, and sometimes I come up on the jump, and perhaps inside of half an hour it will be all I can do to get up at all."

Dr. Willy Meyer was kind enough to excise a piece of the right quadriceps femoris muscle. The results of the microscopic examination are given below.

REMARKS.

There can be no doubt whatever that in this case the diagnosis of Thomsen's disease is correct. In all important points it coincides entirely with the disease as described by Thomsen himself. The only characteristic which is wanting is that of heredity, but it is not possible even here to say that there was no hereditary influence, as the family history is a very unsatisfactory one. The disease in this patient was probably congenital; certainly it has existed as long as he can remember. The myotonic affection, consisting of the peculiar inhibition of motion after a long rest, the slowness and stiffness in executing movements on account of tonic contraction of the muscles after any forced movement, was clearly present. The hardening and contracturing of the muscles after any sudden violent motion; the gradual return to a normal condition after the voluntary movements have been executed several times; the influence of psychical emotion,

of change of temperature, etc., are all perfectly characteristic of the affection. The athletic development of the muscles without any muscular tension during passive motion, and the absence of any other disorder, particularly the absence of any affection of the central nervous system, are all very important. Then, also, the objective examination showed a condition corresponding almost exactly to that found by Erb in his three cases. That the rythmical wave-like contractions, as described by Erb, were not found in this case is unfortunate, as it would have been interesting to corroborate Erb in this particular. Their absence, however, does not in any way invalidate the diagnosis, and it is possible that, if the patient had been under observation for a longer period of time, they might have been seen at some time or other. These undulatory contractions are so interesting that it will not be amiss to cite what Erb says regarding them:

"If a 'large' electrode be placed upon the nape of the neck (or upon the sternum), a 'medium' one upon the palm of the hand, there occurs, with sixteen to eighteen cells, an equable tonic tension of all the muscles of the arm. After short stabile action, or after one or two pole changes, we now see from the lower ends of the flexors of the fingers (above the wrist-joint, volar surface) wave-like contractions take place in rhythmical order; and these wave-like contractions move upwards when the kathode, downwards when the anode is in the hand; therefore, they come from the kathodal side and move towards the anodal side. The single waves follow each other (like the waves of water produced by a falling stone), first with about a second interval (sometimes faster, sometimes slower, this is different on different days), and can be followed up to about the middle of the forearm; then they become slower and disappear after a time. Increase of the current strength may produce them again. The electrodes during this remain perfectly quiet and unmoved." In speaking of this phenomenon as it occurred in his third case, Erb says:

"The best places of application are, for the flexors of the

forearm: palm of the hand or volar surface of the wrist-joint and nape of the neck. For the vastus internus, next to the patella and neck. For the gastrocnemius, tendon of the muscle and neck. The amount of current requisite for the production of the phenomenon varies from six to twenty m.a."

The points in the objective examination which are important are: The long persistence of the mechanical and electrical contractions of the muscles. The slowness and tonic character of the contraction and the difference found between the reactions of muscles and nerves. These differences were briefly as follows: Motor nerves show a normal mechanical excitability. To the faradic current they present qualitatively and quantitively, with the exception of the prolongations of contractions to the strong currents, normal reactions. The galvanic contractions were short and quick, and not prolonged.

The muscles show an exaggerated mechanical excitability. Their galvanic and faradic excitability is quantitively and qualitatively changed. Their contractions are slow, tonic, and prolonged.

For the various changes found in the muscles, Erb proposes the name "myotonic reaction," and says that henceforth all that will be necessary in order to objectively diagnose Thomsen's disease will be a few blows with a percussion hammer and a few closures of the galvanic current with the anode and kathode, and that by this simple examination simulation of this disease can always be detected.

Microscopical Examination of Muscle.—The peculiar electrical and mechanical reactions obtained in the foregoing examination seem to point directly to an affection of the muscles themselves, and indicate the necessity of their careful microscopical examination. Others, also reasoning in same manner, have examined pieces of muscles from myotonia congenita, Ponfick, Petrone, Jacusiel-Grawitz, Knud Pontoppidan, Rieder, and finally, Erb, but all of them, with the exception of Erb, with purely negative results. Erb's microscopical examination of three cases re-

vealed so much that was new that the results in my case, which differ so little clinically from those of Erb, were awaited with considerable interest. I may say right here at the outset that all the points seen by Erb were substantially corroborated by my examination, but still more was seen, and this seems to me, as will be shown, to be of particular importance for the explanation of many of the phenomena seen clinically.

The specimens were obtained from a piece of the quadriceps femoris muscle, which was removed from the pa-

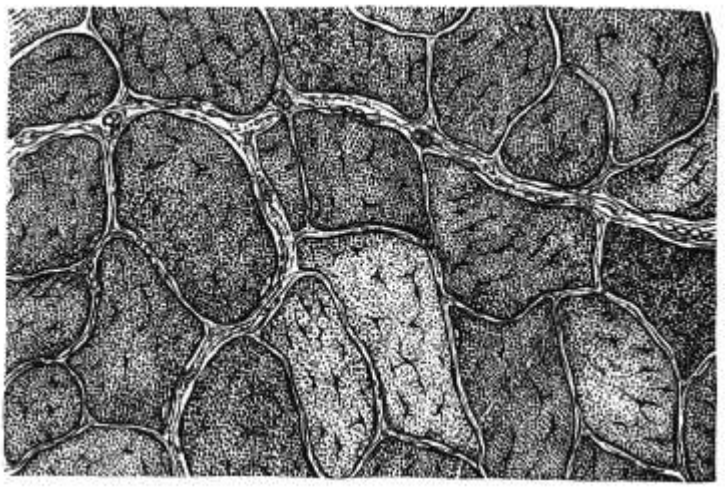

FIG. 1.—Normal muscle. Quadriceps femoris. Transverse section. × 300.

tient under antiseptic precautions, and with the use of an Esmarch bandage. The fresh piece of muscle was dropped immediately after excision into a one-half-per-cent solution of chromic acid and left there until sufficiently hardened. It was then transferred into strong alcohol, imbedded in celloidin and cut both transversely and longitudinally. The sections thus obtained were then stained, some of them in an ammoniacal solution of carmine, and others in a one-half-per-cent solution of chloride of gold. Thereupon the specimens were mounted in glycerin. It is well here to lay stress upon this fact, as the illustrations in Erb's

monograph, repeatedly referred to above, were taken from specimens mounted in Canada balsam, and to this circumstance I am inclined to attribute the lack of details which is apparent in those drawings, and which differs so markedly from that which I have seen. The appearances in my own specimens under a comparatively low power (300 diam.) were so striking that the necessity of comparing them with sections of a normal muscle at once became

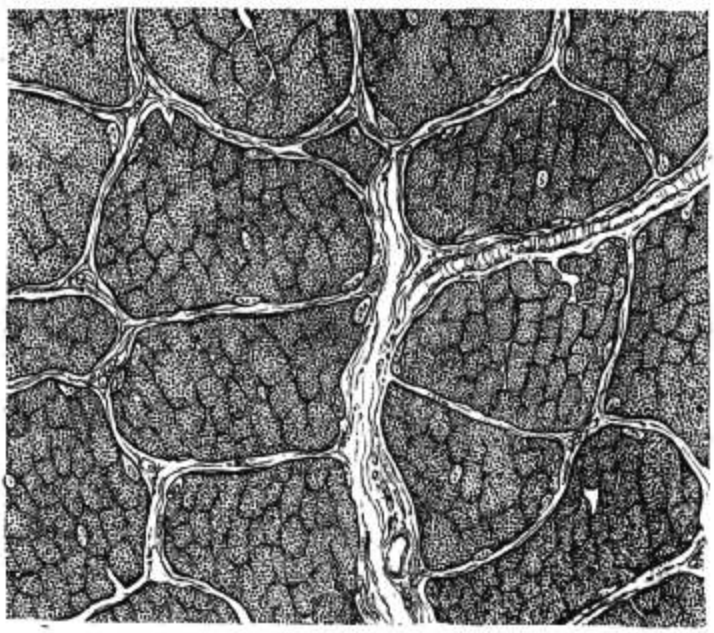

FIG. 2.—Myotonia congenita. Quadriceps femoris. Transverse section. × 300.

apparent. The piece of muscle used for comparison was excised in the same manner, from the same locality, and from a healthy man of nearly the same age and size as was the diseased piece. It was then also placed at once in a one-half-per-cent solution of chromic acid and, in short, treated and mounted in the same manner as described above. Considerable time and care were necessary in order to obtain a suitable subject from whom to take this piece, but this was done purposely, as I feel sure that muscle taken from

a corpse would have been considered, and quite justly I admit, as worthless for the purposes of comparison and of drawing definite conclusions. Transverse sections of the affected muscles revealed with 300 diam. the following facts, substantially in agreement with the statements and conclusions of Erb.

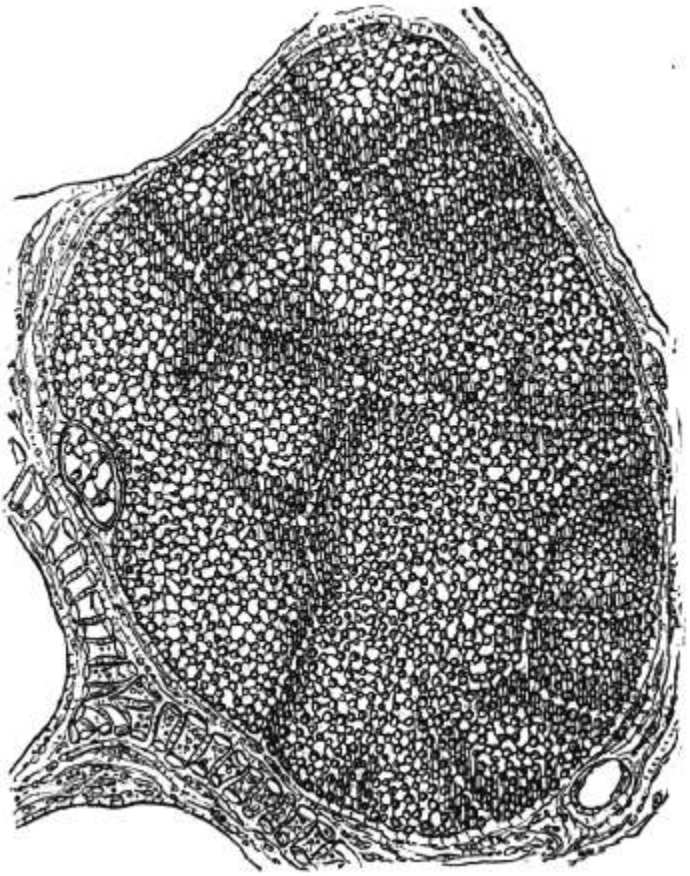

FIG. 3.—Normal muscle-fibre. Quadriceps femoris. Transverse section. × 1200.

1st. The muscle fibres were about double the size of those in normal muscle, or perhaps on an average even larger. The illustrations of Erb's two cases show an increase in the size of the muscle fibres to the threefold or fourfold. My case was evidently, from a microscopical

point of view, not so marked as those of Erb. The fibres were also found to differ in shape from the normal, being more circular with somewhat rounded corners, and not polygonal as if from mutual pressure.

2d. The nuclei of the muscle-fibres were distinctly augmented in number, though also not as markedly as in Erb's cases. At the periphery of some of the fibres entire chains of nuclei could be traced, others showed scattered nuclei even in the middle of the fibre, a feature which is of comparatively rare occurrence in normal muscle.

3d. The connective tissue around the muscle-bundles (external perimysium), and that around the single fibres (internal perimysium), was found distinctly augmented. The internal perimysium, however, not to such a degree as in Erb's cases, since here and there it did not exceed that found in the normal condition. As normally, the external perimysium carried arteries and veins, the internal only capillaries. In several specimens I met with medullated nerve-fibres in the internal perimysium, and have also seen motor end plates at the periphery of some of the fibres, without finding any deviation in the course, number, or terminal arrangements of the nerve apparatus from the normal condition.

The peculiar formation of vacuoles in the muscle-fibres, as seen by Erb in two of his cases, was carefully and repeatedly sought for, but not a single specimen showed anything which could have been taken for them.

Transverse sections of the normal muscle with the same power exhibit marked differences in the interior of the various muscle-fibres. In the first place almost every fibre shows narrow angular slits, sometimes more, sometimes less numerous, which correspond to the embryonic sarcoplasts (Margo), which have entered into the construction of the single fibre. The entire surface of the fibre appears dotted, these dots corresponding, as is well known, to the sarcous elements of Bowman. The dots are either uniformly scattered or are arranged in rows, the latter condition obviously corresponding to a slightly oblique sec-

tion of the fibre. In some fibres also we see the sarcous elements closely packed together and coarse in appearance, in others they are minute, scarcely perceptible, and are widely separated from each other, thus rendering the

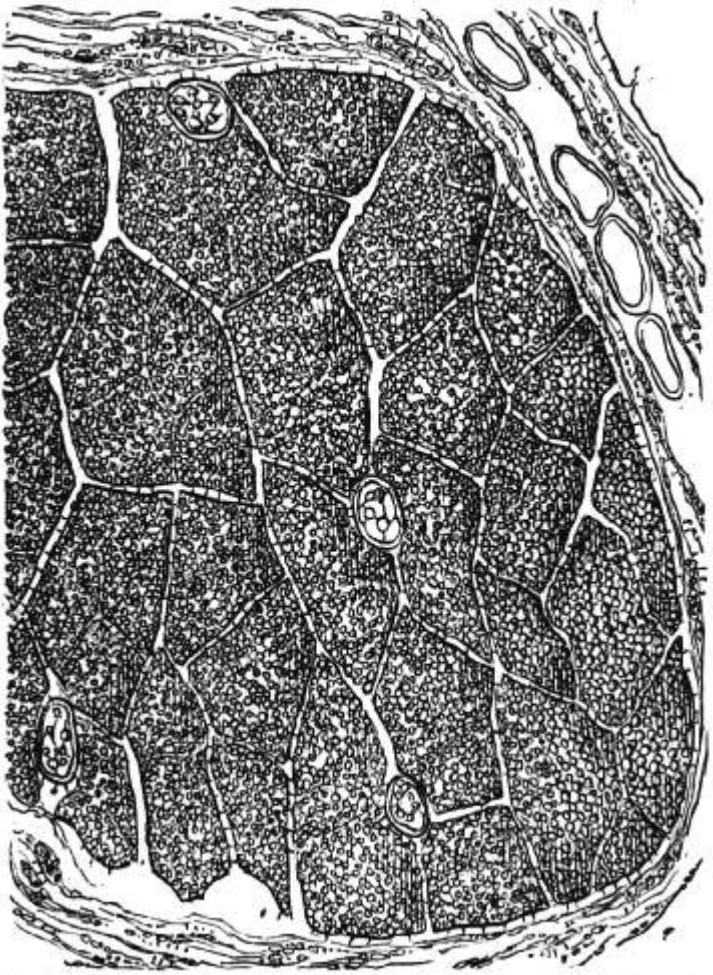

FIG. 4.—Myotonia congenita. Quadriceps femoris. Transverse section of a muscle fibre. × 1200.

appearance of the entire fibre almost homogeneous. We even see these variations in one and the same fibre, it being in part coarsely, in part finely granular. Differences in the arrangement of the sarcous elements are known to exist

in accordance with the degree of contraction or extension of the muscle-fibre. When the muscle is completely at rest, we find the sarcous elements placed at regular intervals and exhibiting a certain size; this, however, varying in different muscles and individuals. During contractions the size of each individual sarcous element will increase and the interstices between them will decrease. In the state of extension, however, the single sarcous elements will decrease in size and the interstices between them become widened.

If now a piece of muscle be removed from the living body and at once placed into a solution of chromic acid, the state of contraction of at least a number of fibres, caused by the abrupt interruption of their continuity and by their exposure to the comparatively cool air, will be fixed by the chromic acid; hence we are justified in expecting to find, at any rate, a number of fibres in this state of contraction. Furthermore, we are justified in expecting to find various degrees of contraction in one and the same fibre. That our expectations are realized practically may be seen by reference to Fig. 1.

The difference in the interior structure of the single muscle-fibres from the case of myotonia congenita, as compared with that of normal muscle, is exceedingly well marked in transverse section. In the first place, nearly every fibre is distinctly divided into angular fields, varying in size, the same as are also seen in normal muscle, where, however, they are but faintly indicated (Cohnheim's fields). This splitting up of the fibre can in some places be traced into extremely minute fields, and in many instances is marked to such a degree that comparatively wide gaps are seen between the angular fields. Here and there also entire fields have dropped out, leaving large gaps behind. This latter feature is not infrequently seen along the periphery of some of the fibres, particularly in such cases where the fibre has been mechanically dragged out from its contiguity with the adjacent fibres. This feature is also alluded to by Erb.

The sarcous elements are also seen to be much more

uniform in size and arrangement than in the normal muscle, this being probably caused by the more general contraction of the muscular fibres (see Fig. 2), or by the uniformly smaller size of the sarcous elements, as mentioned later. Transverse sections of the normal muscle, when viewed with high power (1,200 diam.), show groups of sarcous elements, faintly separated from their neighbors by slight

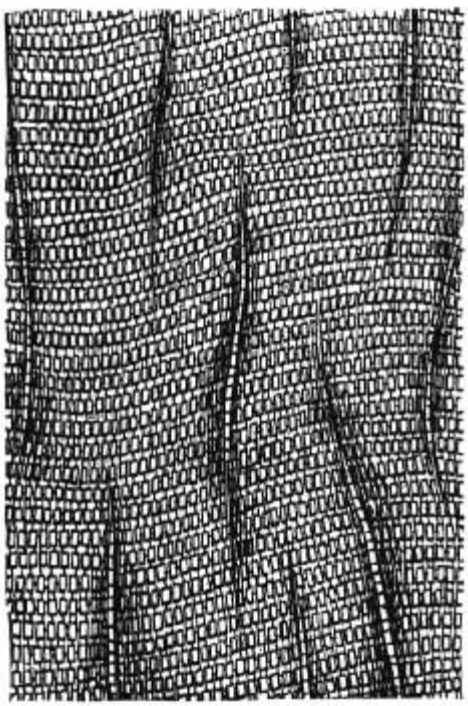

FIG. 5.—Segment of a normal muscle-fibre. Quadriceps femoris. Longitudinal section. × 1200.

interstices. The individual sarcous elements vary slightly in size and, especially in some of the muscle-fibres which apparently are not in a state of intense contraction, are seen to be distinctly connected with the adjacent elements by means of conical threads. These threads also traverse the interstices between the groups of the sarcous elements, and this connection is so well marked that in the entire field of a fibre not a single interruption between in-

dividual sarcous elements and groups of elements is visible.

Muscle-fibres which are evidently in a state of intense contraction show the sarcous elements lying close against each other, so that even when the short threads cannot be made out, a direct continuity is seen to be present by the immediate contact of the sarcous elements between themselves. Whenever a nucleus is seen at the periphery of a fibre, its contour is likewise seen connected with the adjacent sarcous elements by means of these delicate threads.

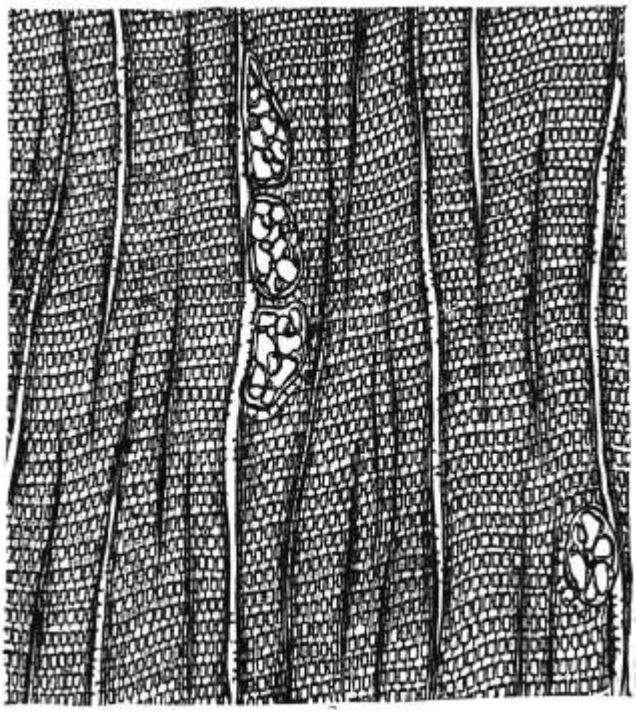

FIG. 6.—Segment of a muscle-fibre from myotonia congenita. Quadriceps femoris. Longitudinal section. × 1200.

Furthermore, wherever there are motor plates at the periphery of a fibre, a continuity is also established between them and the neighboring sarcous elements (Fig. 3). Chloride of gold solution stains the sarcous elements and their connecting threads a violet color, thus rendering them more distinctly visible.

Transverse sections from the muscle affected with myotonia congenita, when regarded with the same power (1,200), show almost everywhere a lack of continuity between the groups of sarcous elements. This feature is most conspicuous along the splits and gaps spoken of above; secondary smaller splits, which traverse the larger groups of sarcous elements, show here and there only a few connecting threads.

The sarcous elements within the smallest fields are closely packed together, so that a mutual contiguity is established without the aid of intervening threads, and signifies, according to the statements made above, a high degree of contraction within a limited number of sarcous elements, and an interruption of continuity between smaller groups of sarcous elements (Fig. 4).

Longitudinal sections of normal muscle, viewed with high power (1,200), show the well-known variations in the arrangement of the rows of sarcous elements, down to the splitting up of the entire fibre into extremely delicate fibrillæ (Schwann's diagram). Within the fibres, slits, as is well known, exist, corresponding to the construction of the entire fibres from a number of embryonic sarcoplasts. Whatever be the arrangement of the rows of sarcous elements, these are invariably seen to be connected with each other by means of delicate threads in a longitudinal as well as in a transverse direction. Such connections are seen even where the sarcous elements form a continuous longitudinal line and the connections are traceable throughout all slits caused by sarcoplasts (see Fig. 5). Longitudinal sections of fibres from the abnormal muscle, with the same power, differ from the normal muscle by the presence of very marked slits, also corresponding to the boundaries of the sarcoplasts, in which, however, no connections are visible.

Particularly in these sections it becomes evident by comparison that all the sarcous elements are markedly smaller in the abnormal than in the normal muscle.

CONCLUSIONS.

Clinically, Thomsen's disease appears to be an affection of the muscles alone, and microscopically this probability is only confirmed. It seems also as if the result of the microscopical examination were capable of casting some light upon the intricate nature of this peculiar affection. There can be no doubt that this disease is a congenital malformation of the muscular fibres, as is proven by the augmented size of each individual fibre, and by the increase in the number of their nuclei and of the perimysium.

It is obvious that a considerably larger number of embryonic sarcoplasts must have entered into the construction of each individual fibre than is the case in normal development. Such a muscle necessarily has a considerably larger number of sarcous elements, or which is synonymous, more contractile matter than a normal muscle, and that, therefore, the contraction of such a muscle is more liable to become exaggerated, as it were; or, in other words, to become tetanic. This is plainly seen in the specimens obtained from my case, where clusters of sarcous elements are seen to aggregate to a close contiguity. In normal muscle, the motor nerves are known to terminate in the form of plates beneath the sarcolemma, but upon the surface of the muscle-fibre. The continuity between the motor end-plate and the adjacent sarcous elements is established by delicate threads, and the continuity throughout the entire muscle-fibre is preserved by such filaments interconnecting all the sarcous elements in every direction, the interstices between the embryonic sarcoplasts not making any exception to this rule. Thus we conceive that the nerve-impulse, whatever this may prove to be, is transmitted from the motor nerve into the terminal plate, thence into the adjacent sarcous elements, and finally into all contractile particles of a muscle in a direct way, namely, by means of the connecting threads. In Thomsen's disease, the motor nerves and motor end-plates do not show any deviations from the normal. The

nerve-impulse, therefore, is transmitted into the muscle-fibre in the same manner as in the normal condition.

The result of this reception of impulse will be a contraction which, especially after a certain rest, will be a hypercontraction, or rather tetanus. This tetanus leads to an agglomeration of a certain number of sarcous elements, with a break in the continuity of the contracted clusters, as can be plainly seen under the microscope. In consequence of this tetanus, the nerve-influence is inhibited for so long as the tetanus lasts. After the lapse of a few seconds, the tetanic contraction will subside, the continuity between the hitherto separated groups of sarcous elements will become re-established, and the propagation of nerve-influence again rendered possible. If now in the light of our microscopical revelations, and in consideration of the above theory, we reconsider the objective symptoms found in Thomsen's disease, we are able to understand the production of a great many of them. That the muscle becomes tetanic under the influence of the will has been explained, that mechanical and electrical stimuli applied to the muscles themselves produce a prolonged contraction is also understood, but how it is that stimuli applied to the nerve do not have the same action as those applied to the muscle is not so clear. This much is certain, that the cause for this variation must be sought in some change in the nerves themselves, and not in the muscles, probably a change in their molecular arrangement, for microscopically the nerve terminations appear normal, and it is after all possible that later observations may discover changes either in the peripheral or central nervous system, which will take this peculiar affection out of the domain of primary muscular disorders to which it now appears to belong.